SMARCAL1 gene

SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a like 1

Normal Function

The *SMARCAL1* gene provides instructions for producing a protein whose specific function is unknown. The SMARCAL1 protein can attach (bind) to chromatin, which is the complex of DNA and protein that packages DNA into chromosomes. Based on the function of similar proteins, the SMARCAL1 protein is thought to influence the activity (expression) of other genes through a process known as chromatin remodeling. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development. When DNA is tightly packed, gene expression is lower than when DNA is loosely packed.

Health Conditions Related to Genetic Changes

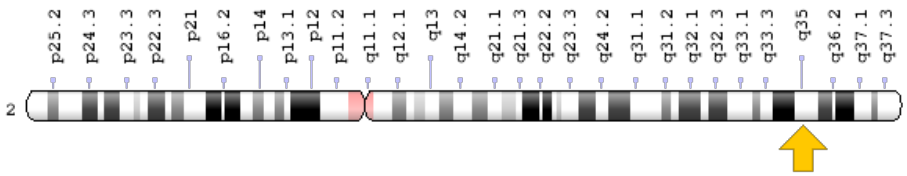
Schimke immuno-osseous dysplasia

More than 40 mutations in the *SMARCAL1* gene have been found to increase the risk of Schimke immuno-osseous dysplasia. The mutations associated with Schimke immuno-osseous dysplasia disrupt the usual functions of the SMARCAL1 protein or prevent the production of any functional protein. People who have mutations that cause a complete lack of functional protein tend to have a more severe form of this disorder than those who have mutations that lead to an active but malfunctioning protein. Mutations in the *SMARCAL1* gene are thought to lead to disease by affecting protein activity, protein stability, or the protein's ability to bind to chromatin. It is not clear how *SMARCAL1* mutations contribute to short stature, kidney disease, and a weakened immune system in people with Schimke immuno-osseous dysplasia. In order for people with *SMARCAL1* gene mutations to develop Schimke immuno-osseous dysplasia, other currently unknown genetic or environmental factors must also be present.

Chromosomal Location

Cytogenetic Location: 2q35, which is the long (q) arm of chromosome 2 at position 35

Molecular Location: base pairs 216,412,414 to 216,483,053 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- HARP
- HepA-related protein
- HHARP
- SMAL1_HUMAN
- SMARCA-like protein 1
- SWI/SNF-related matrix-associated actin-dependent regulator of chromatin a-like 1
- SWI/SNF related, matrix associated, actin dependent regulator of chromatin, subfamily a-like 1

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): ATP-driven Chromatin Remodeling Machines Change Nucleosome Structure https://www.ncbi.nlm.nih.gov/books/NBK26834/#A644

GeneReviews

- Schimke Immunoosseous Dysplasia https://www.ncbi.nlm.nih.gov/books/NBK1376

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SMARCAL1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A-LIKE PROTEIN 1
  http://omim.org/entry/606622

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SMARCAL1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SMARCAL1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11102
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/50485
- UniProt
  http://www.uniprot.org/uniprot/Q9NZC9

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- OMIM: SWI/SNF-RELATED, MATRIX-ASSOCIATED, ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A-LIKE PROTEIN 1
http://omim.org/entry/606622

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/SMARCAL1

Reviewed: November 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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